SERPING1 gene

serpin family G member 1

Normal Function

The *SERPING1* gene provides instructions for making a protein called C1 inhibitor, which is a type of serine protease inhibitor (serpin). Serpins help control several types of chemical reactions by blocking the activity of certain proteins. C1 inhibitor is important for controlling a range of processes involved in maintaining blood vessels, including inflammation. Inflammation is a normal body response to infection, irritation, or other injury.

C1 inhibitor blocks the activity of several proteins in the blood, including plasma kallikrein and the activated form of factor XII (called factor XIIa). These two proteins are involved in the production of bradykinin. Bradykinin is a protein that promotes inflammation by increasing the permeability of blood vessel walls, allowing fluids to leak into body tissues. C1 inhibitor attaches (binds) to plasma kallikrein and factor XIIa, which prevents them from completing any further reactions. These proteins are cleared from the bloodstream once they are bound to C1 inhibitor.

Health Conditions Related to Genetic Changes

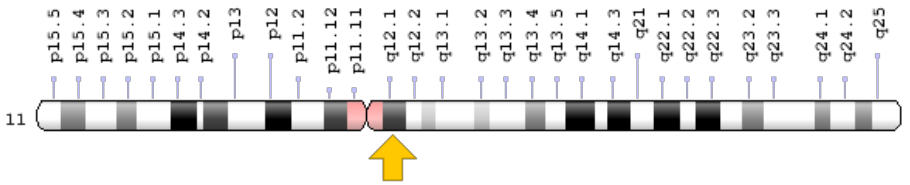
hereditary angioedema

More than 250 mutations in the *SERPING1* gene have been found to cause hereditary angioedema types I and II. Mutations that cause type I occur throughout the gene and lead to reduced levels of C1 inhibitor in the blood. Mutations that cause type II usually occur in a specific region of the gene called exon 8 and result in the production of a C1 inhibitor that functions abnormally. Without the proper levels of functional C1 inhibitor, the activity of plasma kallikrein and factor XIIa cannot be blocked and excessive amounts of bradykinin are produced. Excess fluids leak through blood vessel walls and accumulate in body tissues, leading to the recurrent episodes of swelling seen in individuals with hereditary angioedema type I and type II.

Chromosomal Location

Cytogenetic Location: 11q12.1, which is the long (q) arm of chromosome 11 at position 12.1

Molecular Location: base pairs 57,597,554 to 57,614,853 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- C1-INH
- C1IN
- C1INH
- C1NH
- complement component 1 inhibitor
- IC1_HUMAN
- plasma protease C1 inhibitor
- serine/cysteine proteinase inhibitor clade G member 1
- serpin peptidase inhibitor, clade G (C1 inhibitor), member 1

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SERPING1%5BTIAB%5D%29+OR+%28%28C1IN%5BTIAB%5D%29+OR+%28C1NH%5BTIAB%5D%29+OR+%28HAE1%5BTIAB%5D%29+OR+%28HAE2%5BTIAB%5D%29+OR+%28C1INH%5BTIAB%5D%29+OR+%28complement+component+1+inhibitor%5BTIAB%5D%29+OR+%28plasma+protease+C1+inhibitor%5BTIAB%5D%29+OR+%28C1-INH%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- COMPLEMENT COMPONENT 1 INHIBITOR
  http://omim.org/entry/606860

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SERPING1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SERPING1%5Bgene%5D
- HGNC Gene Family: Serpin peptidase inhibitors
  http://www.genenames.org/cgi-bin/genefamilies/set/739
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1228
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/710
- UniProt
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